

First-trimester ultrasonography revealed a gestational sac featuring cystic spaces and no visible embryo: a case of trisomy 7

İlk trimester ultrasonografide kistik alanlar içeren, embriyo görülmeyen gebelik kesesi: Bir trizomi 7 olgusu

Aysel Derbent¹, Zerrin Yılmaz², Serap Simavlı¹, Özge Özer², Nilgün Öztürk Turhan¹

¹Department of Obstetrics and Gynecology, Fatih University, Ankara, Turkey

²Department of Medical Genetics, Başkent University, Ankara, Turkey

Abstract

Ultrasound examination in early pregnancy has steadily gained importance and is now routine for most women in the first trimester. The sonographic features of early trisomy 7 pregnancies are not well characterized.

We present a case of trisomy 7 in which early pregnancy ultrasound revealed a gestational sac featuring cystic spaces and no visible embryo.

Based on comparison with a previously reported case of trisomy 7 featuring a multicystic anembryonic gestational sac we suggest that this ultrasonographic finding may be a sign of trisomy 7.

(J Turkish-German Gynecol Assoc 2010; 11: 61-2)

Key words: Trisomy 7, ultrasonography, early pregnancy failure

Received: 17 June, 2009

Accepted: 3 September, 2009

Özet

Erken gebelikte ultrasonografi muayenesi gittikçe önem kazanmıştır ve şu anda birçok gebeliğin birinci trimesterinde rutin uygulanmaktadır. Trizomi 7 olgularının erken ultrasonografik özellikleri tam belirlenmemiştir. Burada erken gebelik ultrasonografisinde kistik alanlar olan bir gebelik kesesinde embriyonun görülmediği bir trizomi 7 olgusunu sunuyoruz.

Daha önce rapor edilmiş olan bir trizomi 7 olgusundaki multikistik anembryonik gestasyonel kese tanısını da göz önüne alarak bu ultrasonografik bulgunun trizomi 7 belirtici olabileceğini öneriyoruz.

(J Turkish-German Gynecol Assoc 2010; 11: 61-2)

Anahtar kelimeler: Trizomi 7, ultrasonografi, erken gebelik kaybı

Geliş Tarihi: 17 Haziran 2009

Kabul Tarihi: 03 Eylül 2009

Introduction

Ultrasound examination in early pregnancy is now routine for most women in the first trimester. Determination of whether some specific karyotype abnormalities are linked with distinct ultrasonographic findings is gaining importance.

Case report

A 23-year-old primigravida woman was admitted to the obstetrics unit of our hospital at 7 weeks 6 days of gestation with the complaint of vaginal bleeding. The woman and her husband were both healthy, the marriage was not consanguineous, and there was no family history of congenital malformations. Six days prior to presentation, the patient had been examined at another hospital for menstrual delay. At that time her blood beta-human chorionic gonadotropin (β -hCG) level was 9740 IU/mL. Transvaginal ultrasound showed a single intrauterine gestational sac containing a yolk sac but no fetal pole. Our ultrasonographic examination at 7

weeks 6 days of gestation showed a large gestational sac of mean diameter 38 mm, three cystic spaces in the chorionic sac (one cyst inside another, and a third tangent to these), and no visible embryo (Figure 1). The chorion appeared normal. There was no solid component in the gestational sac. The patient was diagnosed with anembryonic pregnancy. Two days later the pregnancy was terminated using the Carmen aspiration method, and two samples of the evacuated tissues were collected: one that appeared to be chorionic villi (specimen 1) and one that resembled endometrium (specimen 2). These were sent for chromosome analysis. Two weeks after termination, ultrasonography showed a normal central endometrial echo and blood testing revealed β -hCG of 3 IU/mL. The karyotyping results were 47,XX,+7 (specimen 1) (Figure 2) and 46,XX (specimen 2). The family refused any further investigation and it was thus impossible to perform molecular analyses to rule out maternal contamination. During genetic counseling, the parents were informed about the detected abnormality and prenatal diagnostic tests were recommended for any future pregnancies.

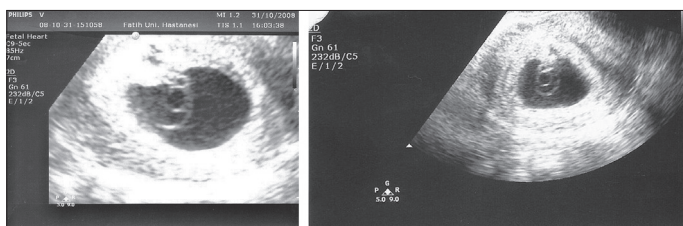


Figure 1. Ultrasonographic examination at 7 weeks 6 days of gestation showed a large gestational sac, three cystic spaces inside the chorionic sac, and no visible embryo

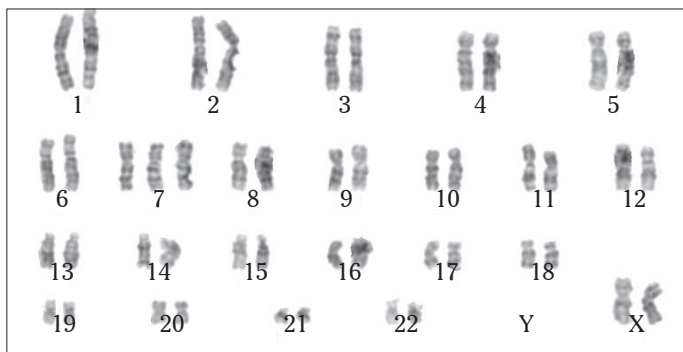


Figure 2. Representative karyotype from specimen 1, showing complete trisomy of chromosome 7

Discussion

Research indicates that nearly 40% of early pregnancies result in miscarriage, and many authors have investigated the role of chromosomal abnormalities in these embryonic losses (1). One such study looked at 144 spontaneous abortions through direct sampling of chorionic villi (2). The authors found that 100 (70%) of these specimens had abnormal chromosomes, and 64% of those 100 were autosomal trisomies. Two trials examined the correlation between karyotype and ultrasound findings in patients with failed early pregnancy. Goldstein et al. studied 102 women with ultrasound diagnosis of early pregnancy failure and found that 44 of these pregnancies (43%) featured abnormal karyotypes (3). Thirty-three (75%) of the 44 were trisomy cases. Coulam et al. examined 137 spontaneous abortions and found 86 (63%) cases with abnormal karyotypes (68 aneuploidies and 18 polyploidies) (4). Neither of these papers reported a case of trisomy 7. Both sets of authors concluded that ultrasonographic findings cannot predict karyotype in cases of spontaneous abortion, but they called for further studies to determine whether some specific karyotype abnormalities are linked with distinct ultrasonographic findings.

The second specimen from our patient appeared to be endometrium, and the karyotype for this tissue was 46 XX. Examination of tissue from curettage for reliable separation of chorionic villi from decidua was initially proposed on a clinical basis. This method allows a portion of chorionic villi to be distinguished from maternal decidua and was submitted separately for chromosomal analysis (3).

Four percent to 10% of all trisomy cases are trisomy 7, and this abnormality is generally considered to be lethal during embryogenesis. Trisomy 7 is usually detected by chorionic villus sampling due to confined placental mosaicism in an

ultrasonographically normal pregnancy and the outcome is normal, without intrauterine fetal growth retardation (5). Almost all surviving children are mosaics and exhibit variable and nonspecific clinical features. A patient with typical Potter syndrome and full trisomy 7 was described in 1980 by Yunis et al. (6). Biri and colleagues presented a case of double aneuploidy, namely, trisomy 7 and X mosaicism, with characteristic features of Potter syndrome (7). The fetus survived in utero until the 32nd week of gestation. The same authors also noted four previously reported cases of trisomy 7 that survived into the third trimester and all of these exhibited features indicative of the Potter sequence.

The sonographic features of early trisomy 7 pregnancies are not well characterized. A case similar to ours was reported in 2001 by Ojha et al. (8). In that instance, ultrasonography at 8 weeks and 1 day gestation showed a heterogeneous multicystic mass with no visible embryo, and karyotyping revealed trisomy 7. The cystic appearance of that abnormal pregnancy is almost identical to what we observed in our patient. Our literature search revealed no other reports of the sonographic features of full trisomy 7 pregnancies. Based on this comparison with a previously reported case of trisomy 7 featuring a multicystic anembryonic gestational sac, we suggest that this ultrasonographic finding may be a sign of trisomy 7.

Women and couples who experience failed pregnancy need more detailed information about the reasons for the failure. In each case it is essential to assess the various causes of miscarriage. Detection of a chromosomal aberration provides a definitive diagnosis and eliminates the need for any further investigation. The ability to identify specific ultrasonographic findings that may predict abnormal karyotypes is extremely valuable and is gaining importance. Our case suggests that there may, indeed, be certain characteristic features of trisomy 7 detectable on ultrasound in early pregnancy.

Conflict of interest

None declared

References

1. Regan L, Braude PR, Trembath PL. Influence of past reproductive performance on risk of spontaneous abortion. *Br Med J* 1989; 26: 541-5.
2. Ohno M, Michiko BS, Maeda T, Matsunobu A, Akira MD. A cytogenetic study of spontaneous abortions with direct analysis of chorionic villi. *Obstet Gynecol* 1991; 77: 394-8.
3. Goldstein SR, Kerenyi T, Scher J, Papp C. Correlation between karyotype and ultrasound findings in patients with failed early pregnancy. *Ultrasound Obstet Gynecol* 1996; 8: 314-7.
4. Coulam CB, Goodman C, Dorfmann A. Comparison of ultrasonographic findings in spontaneous abortions with normal and abnormal karyotypes. *Human Reproduction* 1997; 12: 823-6.
5. Chromosomal syndromes: unusual variants; In Gorlin RJ, Cohen MM Jr, Hennekam RCM (eds): *Syndromes of the head and neck*. Fourth edition. Oxford: Oxford University Press, Inc 2002, pp 76-91.
6. Yunis E, Ramirez E, Uribe JG. Full trisomy 7 and Potter syndrome. *Hum Genet* 1980; 54: 13-8.
7. Biri A, Karaoğuz MY, Ince GD, Ergün MA, Menevşe S, Bingöl B. Double aneuploidy involving trisomy 7 with Potter sequence. *Eur J Med Genet*. 2005; 48: 67-73.
8. Ojha K, Sladkevicius P, Parikh B, Moscoso G, Nargund G. Trisomy 7 following assisted conception treatment. *Ultrasound Obstet Gynecol* 2001; 17: 543-5.